Claims

1 A method for the diagnosis of a polymorphism in a PDH E1α gene in a human, which method comprises determining the sequence of the nucleic acid of the human at position 1388 in the PDH E1α gene as defined by the position in SEQ ID NO: 2, and/or at one or more of positions 26 and 161 of intron 7 of the PDH E1α gene as defined in SEQ ID NO.1; and determining the status of the human by reference to polymorphism in the PDH E1α gene.

2 A method according to claim 1 in which the polymorphisms are further defined as:

Position	Reference	Region	Polymorphism
26	SEQ ID NO:1	intron 7	(GGCCAA)n
161	SEQ ID NO:1	intron 7	C/A
1388	SEQ ID NO:2	3' UTR	C/T

3 A method according to claim 2 which comprises diagnosis of the following haplotype:

Position	Reference	Region	Polymorphism
26	SEQ ID NO:1	intron 7	$(GGCCAA)_2$
161	SEQ ID NO:1	intron 7	A

4 A nucleic acid comprising the nucleic acid of SEQ ID NO.1 or a sequence at least 85% homologous thereto; or a complementary strand thereof or an antisense sequence thereto or a fragment thereof of at least 20 bases comprising at least one positions 26 or 161.

5 An allele specific primer capable of detecting a PDH E1α gene polymorphism at one or more of position 1388 in the PDH E1α gene as defined by the position in SEQ ID NO: 2 and/or positions 26 and 161 of intron 7 of the PDH E1α gene as defined by the positions in SEQ ID NO.1.

6 An allele-specific oligonucleotide probe capable of detecting a PDH E1α gene polymorphism at one or more of position 1388 in the PDH E1α gene as defined by the position in SEQ ID NO: 2 and/or positions 26 and 161 of intron 7 of the PDH E1α gene as defined by the positions in SEQ ID NO.1.

7 Use of any polymorphism as defined in claim 2 as a genetic marker in a linkage study.

position in SEQ ID NO: 2 and/or positions 26 and 161 of intron 7 of the PDH E1α gene as defined by the positions in SEQ ID NO.1.

7 Use of any polymorphism as defined in claim 2 as a genetic marker in a linkage study.

8 A method of treating a human in need of treatment with a PDH drug in which the method comprises:

i) diagnosis of a polymorphism in the PDH E1α gene in the human, which diagnosis comprises determining the sequence of the nucleic acid at one or more of position 1388 in the PDH E1α gene as defined by the position in SEQ ID NO: 2 and/or positions 26 and 161 of intron 7 of the PDH E1α gene as defined by the positions in SEQ ID NO.1, and determining the status of the human by reference to polymorphism in the PDH E1α gene; and

ii) administering an effective amount of a PDH drug.

9 Use of any one of the following in bioinformatic analysis:

i) any polymorphism as defined in claim 1 or 2;

ii) the haplotype defined in claim 3; or

iii) a nucleic acid sequence as defined in claim 4.

10 A use according to claim 9 comprising a bioinformatic analysis selected from homology searching, mapping, haplotyping, genotyping or pharmacogenetic analysis.